

# Genetics

## Newborn screening for sickle cell anemia

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Early identification of sickle cell anemia (SCA) with newborn screening enables antibiotic prophylaxis for *Streptococcus pneumoniae*, which is one of the early life-threatening complications of untreated disease. Identifying newborns with SCA also facilitates the education of parents and caregivers about early treatment of crises and prevention of complications. Newborn screening might also detect other hemoglobin (Hb) variants in children, including HbC, HbD, and HbE; however, newborn screening does not test reliably for other hemoglobinopathies, such as  $\beta$ -thalassemia or HbH disease. Hemoglobinopathy screening should therefore still be offered to those parents from high-risk ethnic backgrounds, ideally before or early in pregnancy.

**Bottom line.** In some provinces, newborn screening for SCA is available, facilitating early interventions to prevent complications.

The complete *Gene Messenger—Newborn Screening for Sickle Cell Anemia* by the GenetiKit research team is available on **CFPlus**.<sup>\*</sup> Past Gene Messenger articles can be accessed on-line at [www.cfp.ca](http://www.cfp.ca). On the home page, click on **Collections** in the left-hand menu, then click on **Genetics**.

### Competing interests

None declared

The GenetiKit research team, a group of family physicians, genetic counselors and geneticists, designed the Gene Messenger series to provide practical information to help family physicians and their patients make informed choices about rapidly emerging genetic discoveries. The series is a collection of up-to-date, definitive, short reviews on genetics topics that have made headlines, and offers recommendations regarding referral for genetic services or testing.

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## GENE MESSENGER

For more information on genetics topics,  
 see [www.mtsinai.on.ca/FamMedGen/](http://www.mtsinai.on.ca/FamMedGen/)



<sup>\*</sup>The Gene Messenger on newborn screening for sickle cell anemia is available at [www.cfp.ca](http://www.cfp.ca). Go to the full text of this article on-line, then click on **CFPlus** in the menu at the top right-hand side of the page.



# Dermacase



## Can you identify this condition?

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A 30-year-old man presents with monomorphic, discrete, smooth, dome-shaped, skin-coloured papules of 1 to 2 mm in length on the corona and sulcus of the glans penis. The lesions are asymptomatic and he thinks that they have increased in number over the past year. His medical history is unremarkable and he is not taking any medications. He is sexually active and occasionally wears condoms.

### The most likely diagnosis is

1. Condyloma acuminata
2. Lichen nitidus
3. Pearly penile papules
4. Ectopic sebaceous glands
5. Molluscum contagiosum

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### 3. Pearly penile papules

Pearly penile papules (PPPs) are benign angiofibromas that present as a single row or several rows of discrete, smooth, dome-shaped, 1- to 2-mm sized, skin-coloured papules distributed circumferentially on the corona and sulcus of the glans penis.<sup>1</sup> The penile shaft is rarely involved. Although PPPs are asymptomatic, patients often seek treatment because of concerns about cosmetic appearance or concerns about having a sexually transmitted infection, especially genital warts.<sup>2</sup>

Pearly penile papules develop most commonly in the second and third decades of life and less frequently after the age of 40. In men younger than 50 years of age, the prevalence is about 35%.<sup>3</sup> A higher incidence of PPPs has been noted in black and uncircumcised male patients.<sup>4</sup>

Pearly penile papules are characterized histologically as acral angiofibromas. They are thought to represent phylogenetic residua from animal ancestry and are thus considered a normal anatomic variant of the penis.<sup>5</sup> A specific function has not been identified. It was previously suggested that the human papillomavirus might play a role in the etiology of PPPs; however, such an association has not been confirmed.<sup>3</sup> The development of malignancy from PPPs has not been reported.

### Diagnosis

A diagnosis of PPPs is made clinically. The differential diagnosis includes condyloma acuminata (genital warts), molluscum contagiosum, ectopic sebaceous glands, and lichen nitidus.<sup>4</sup> Condyloma acuminata are most important to rule out. Unlike PPPs, they are less uniform in shape and size, tend to be more verrucous, and might change in appearance over time. In addition, they are not typically arranged in neat circular rows surrounding the penis, as are PPPs.<sup>4</sup> Lesions of molluscum contagiosum tend to be umbilicated and larger in size, sometimes with a pink hue.<sup>1</sup> Ectopic sebaceous glands and lichen nitidus are both commonly located on the penile shaft. Ectopic sebaceous glands are more yellow in colour and can often discharge a cheesy material.<sup>4</sup> Lesions of lichen nitidus tend to be flat-topped and are often polygonal in shape.<sup>1</sup> In cases in which the diagnosis is uncertain, a biopsy can be performed, as PPPs and each of the conditions considered in the differential diagnosis have characteristic histologic findings.

### Treatment

As PPPs are benign, no treatment is required. Patients should be reassured that PPPs are not due to a sexually transmitted infection and do not carry any risk of becoming malignant. For those patients with ongoing cosmetic concerns resulting in psychological distress, treatment can be implemented. Cryotherapy with liquid nitrogen and carbon dioxide laser ablation therapy are the 2 most effective treatment modalities. Two treatments with liquid

nitrogen can successfully remove 80% to 90% of lesions without scarring or postinflammatory pigment changes if performed by an experienced physician.<sup>6,7</sup> Excellent cosmetic results have also been obtained with carbon dioxide laser ablation therapy,<sup>8-10</sup> with the continuous wave laser being more effective than the pulsed laser.<sup>10</sup> Pearly penile papules can also be treated successfully with electrodesiccation and simple surgical excision.<sup>4</sup>

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#### Competing interests

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